Claims

1. A method of mutation analysis of a target nucleic acid, said method comprising:

incubating a sample comprising said target nucleic acid in a reaction mixture, in the presence of at least one first nucleic acid and at least one second nucleic acid, wherein said first nucleic acid comprises a primer sequence which anneals to a unique site of a sequence of SEQ ID NO. 1 or 2, and said second nucleic acid has an opposite orientation from said first nucleic acid, and wherein said incubation produces amplified products;

generating duplexes in said amplified products; and

detecting the presence or absence of a heteroduplex from said duplexes, wherein the presence of a heteroduplex indicates the presence of a potential mutation in said target nucleic acid, and wherein the absence of a heteroduplex indicates the absence of a mutation in said target nucleic acid.

2. The method of claim 1, the method further comprising

determining the sequence of a heteroduplex region; and comparing the sequence of the heteroduplex region to SEQ ID NO. 1 or 2; wherein a sequence difference in the heteroduplex region compared to SEQ ID NO. 1 or 2 resulting in a predicted functional change in the protein encoded by said target nucleic acid is indicative of a mutation in said target nucleic acid.

3. The method of claim 1, wherein said first or second nucleic acid comprises a sequence selected from the group consisting of SEQ ID NOs. 3-49.

4. The method of claim 1, said method further comprising performing a nested amplification reaction using said amplified products generated by said first and second nucleic acids as templates and generating duplexes in amplified products from said nested amplification.

5. The method of claim 4, wherein said nested amplification reaction is performed using at least one primer selected from the group consisting of SEQ ID NOs. 3-49 and their complementary sequences.

6. The method of claim 1, wherein identifying the presence or absence of a heteroduplex from said duplexes is performed by DHPLC.

7. The method of claim 1, wherein the sequence of the heteroduplex region is determined by DNA sequencing.

8. The method of claim 1, wherein said second nucleic acid comprises a primer sequence which anneals to a unique site within a sequence of SEQ ID NO. 1 or 2.

9. The method of claim 1, wherein said sample comprising said target template is selected from the group consisting of: genomic DNA, cDNA, total RNA, mRNA, and a cell sample.

10. The method of claim 1, wherein said incubating comprises an amplification reaction selected from the group consisting of: a polymerase chain reaction, a ligase chain reaction (LCR) and a nucleic acid-specific based amplification.

11. The method of claim 1, further comprising confirming the amplified product is a PKD-specific product with one or more restriction enzymes.

12. The method of claim 11, wherein said restriction enzyme cleaves a PKD-specific product to generate a digestion pattern distinguishable from a PKD homologue product.

13. The method of claim 11, wherein said restriction enzyme is selected from the group consisting of: Pst I, Stu I, Xma I, Mlu I, Pvu II, BssHII, Fsp I, Msc I, and Bln I.

14. A diagnosis method for identifying a patient affected with PKD, said method comprising:

(c) obtaining a sample from an individual;

(d) incubating said sample in a reaction mixture, in the presence of at least one first nucleic acid and at least one second nucleic acid, wherein said first nucleic acid comprises a primer sequence which anneals to a unique site within a sequence of SEQ ID NO. 1 or 2, and said second nucleic acid has an opposite orientation from said first nucleic acid, and wherein said incubation produces amplified products;

(c) generating duplexes in said amplified products;

(d) detecting the presence or absence of a heteroduplex from said duplexes, and(e) determining the sequence of the heteroduplex region wherein the presence of a mutation in the heteroduplex region as compared to SEQ ID No. 1 or 2 is indicative that said individual is affected with PKD.

15. The method of claim 14, wherein said detection of a hereroduplex is performed by DHPLC.

16. The method of claim 14, wherein said sequence is determined by DNA sequencing.

17. The method of claim 14, wherein said second nucleic acid comprises a primer sequence which anneals to a unique site within a sequence of SEQ ID NO. 1 or 2.

18. The method of claim 14, wherein said first or second nucleic acid comprises a primer sequence selected from the group consisting of SEQ ID NOs. 3-49.

19. The method of claim 14, said method further comprising performing a nested amplification reaction using said amplified products generated by said first and second nucleic acids as templates and generating duplexes from said nested amplification.

20. The method of claim 19, wherein said nested amplification reaction is performed using at least one primer selected from the group consisting of SEQ ID NOs. 3-49 and their complementary sequences.

21. The method of claim 14, wherein said sample is selected from the group consisting of: a genomic DNA, cDNA, total RNA, mRNA, and a cell.

22. The method of claim 14, wherein said amplification reaction is selected from the group consisting of: a polymerase chain reaction, a ligase chain reaction (LCR) and a nucleic acid-specific based amplification.

23. The method of claim 14, further comprising verifying a said specifically amplified product with one or more restriction enzymes.

24. The method of claim 23, wherein said restriction enzyme cleaves a PKD-specific product to generate a digestion pattern distinguishable from a PKD homologue product.

25. The method of claim 24 wherein said restriction enzyme is selected from the group consisting of: Pst I, Stu I, Xma I, Mlu I, Pvu II, BssHII, Fsp I, Msc I, and Bln I.